Supplementary Figure 3. Pedigree structure, haplotype analyses and conservation of the SHANK3 mutations and variants identified in individuals with autism. In family ASD 1, the proband carries a de novo 22q13 deletion on the paternal chromosome. In family ASD 2, the two affected siblings carry a G insertion on the maternal chromosome, originating from a germline mosaicism. The insertion in exon 21 of SHANK3 leads to a premature truncated protein. The proband of family ASD 4 carries the R12C SHANK3 mutation, transmitted by the mother and shared with his healthy brother. The study of 10 SNPs revealed that the two brothers carrying the R12C variation don’t share the same paternal allele of SHANK3. The proband of family ASD 5 carries the R300C SHANK3 mutation, transmitted by the mother, located in the ankyrin domain. The promoter region, the 5’UTR and the 3’UTR of SHANK3 were sequenced in the patients ASD 4 and ASD 5, but no additional variations were identified.